

Obstetrics and Gynaecology - Women's and Children's Services

Antenatal Screening Result for Down's Syndrome, Edwards' Syndrome and Patau's Syndrome

Insert Banda Label

Your recent screening test to show your chances of having a baby with Down's Syndrome has come back giving you a chance of at term and Edwards' or Patau's Syndrome a chance of at term.

If you have an increased chance of having a baby with Down's, Edwards' or Patau's Syndrome you will be offered a diagnostic test. This can either be a CVS or an Amniocentesis depending on the stage of your pregnancy and the position of your placenta in the womb. For most women the laboratory test will give a definite 'yes' or 'no' answer.

What is a CVS?

While having an ultrasound scan to check the position of the baby and placenta, a fine needle is inserted through the abdomen into the placenta to remove a small amount of placental tissue. This test looks at your baby's chromosomes in the placental tissue. It is most safely performed between the 11th week of pregnancy and the end of the 13th week, but it is occasionally offered later in pregnancy if there is a specific clinical need.



What is an Amniocentesis?

While having an ultrasound scan to check the position of the baby, a fine needle is inserted through the abdomen into the womb to withdraw a small amount of amniotic fluid (fluid in the womb surrounding the baby). This fluid contains cells from the baby which will be examined in the laboratory to look at your baby's chromosomes. This test is usually offered from 15 weeks.

What are Chromosomes?

Chromosomes are the packages in our cells that make up each person's unique genetic blueprint. There are normally 46 in each cell and can be matched up into 23 pairs. Down's Syndrome is a 'trisomy' of chromosome 21; this means three chromosomes on chromosome number 21. Edwards' Syndrome is a trisomy of chromosome 18, and Patau's Syndrome is a trisomy of chromosome 13.

What are the risks with the procedures?

There is a risk of miscarriage with CVS and amniocentesis.

The risk of miscarriage for both tests is greatest during the first 48 hours but continues for about 2 weeks. We advise women to avoid high impact activity for at least the first two days after a procedure.

Occasionally it is not possible to perform the test or obtain enough of a sample during the test. If this happens you will be given the option of coming back at another time.

Occasionally the laboratory is not able to get a result from the sample. You will then be given the option of having the test repeated.

Occasionally the baby may have some cells with chromosomes that are affected with Down's, Edwards' or Patau's Syndrome and some cells that are not affected. This form of a condition is called mosaicism. Very occasionally a test can fail to pick up this form of a Syndrome.

Medication

Certain medication needs to be temporarily suspended prior to an invasive test, e.g. anticoagulant therapy. All current medications will be discussed prior to appointments being confirmed for invasive procedures.

How do I get my results?

The result takes three to five working days. Routinely, the report will include the results for the three most common trisomies. Trisomy of chromosome 21 (T21) being Down's Syndrome; trisomy of chromosome 18 (T18) being Edwards' Syndrome; and trisomy of chromosome 13 (T13) being Patau's Syndrome.

If there are indications for further investigations, these will be requested by your Fetal Medicine clinician. These results will be reported as soon as they are available. You will receive a phone call from the Screening Midwife to inform you of your results.

Do I have to have a diagnostic test?

No; this decision is yours, based on the information given to you about your screening results and the information about the diagnostic tests. You may decide that your chance of having a baby with Down's, Edwards' or Patau's Syndrome is more acceptable than the chances of having an invasive test. The care you receive throughout your pregnancy will carry on as normal. The NHS does not offer any other tests that will give you a definite answer for Down's Syndrome.

What about a scan?

A scan cannot detect whether or not the baby has Down's Syndrome. Edwards' and Patau's Syndromes are checked for on the routine anomaly scan, between 18+0 – 20+6 weeks, though are not always identified.

More detailed information can be found in the booklet, Screening Tests for You and Your Baby, which you will have been given by your community midwife.

Patient Information

If you decide to have a CVS or amniocentesis, please bring your Patient Held Records with you when you come for the test. You can have one other adult with you during the procedure but children are not permitted.

If you have any questions or concerns please phone one of the numbers below:

Antenatal Screening Midwives: 024 7696 7412

Fetal Medicine Department: 024 7696 6572

Scan Department: 024 7696 7348

Antenatal Clinic: 024 7696 7350

Ward 23 (Gynaecology): 024 7696 7000

Phone this number if you get any pain, bleeding, fluid loss, or feel feverish, following your amniocentesis or CVS.

The Trust has access to interpreting and translation services. If you need this information in another language or format please contact the Antenatal Clinic on 024 7696 7350 and we will do our best to meet your needs.

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